Familial Hyperinsulinism (ABCC8-Related)

ABCC8-related familial hyperinsulinism is a very rare condition. It is characterized by very high levels of insulin production. The levels are so high that it can cause low blood sugar levels, and from damage to the pancreas. People with ABCC8-related familial hyperinsulinism are often those who have two variants in the ABCC8 gene.

play+dfe27e1513, you could have one of the variants we tested.

You could pass the variant on to your children.

We could not determine your result for one variant. This can be caused by random test error, other factors that interact with the test, or if you have a copy of a variant on each arm.

How To Use This Test

This test does not diagnose any health conditions.

You should talk to a healthcare professional if this condition or your family's history might affect your children, or if you are concerned about your results.

Important Ethics

This test is most relevant for people of Ashkenazi Jewish descent.

Your results may be relevant if you or your partner are both carriers, such that if you have a 25% chance of passing the condition. If you are a carrier, even with a partner who is not a carrier, you have a 50% chance of passing the condition to your children. If both parents are carriers, you and your partner are both carriers, and you have a 75% chance of passing the condition to your children.

If you and your partner are both carriers, this might have a 25% chance of having a child with the condition. If you have a 25% chance of passing the condition, it might be helpful to talk to your partner to make a decision about whether you want to have children. If you are a carrier and your partner is not a carrier, you have a 50% chance of passing the condition to your children. This might be helpful to talk to your partner about whether you want to consider testing if you plan to have children.

About Familial Hyperinsulinism

Also known as: Congenital Hyperinsulinism, Persistent Hyperinsulinism Hyperglycemia of Infancy (PHHI)

When it develops

Symptoms typically develop the day after birth or in early childhood.

Signs and symptoms

• High levels of insulin
• Low blood sugar
• Low energy
• Irregularities
• Seizures
• Brain damage

Read more at: Genetics Home Reference® "Sudden unexplained death in infancy"

How to manage

Ethics most affected

This condition is most common in people of Jewish ancestry. If your partner is also a carrier, it might be helpful to talk to a healthcare professional about the implications of this condition.

How it is treated

There is currently no cure for this condition. Treatment depends on the severity of the condition. Some people can maintain daily blood sugar levels through medication or diet. Other people may require surgery to remove part of the pancreas.

Consider talking to a healthcare professional if you are thinking about having children.

If you are thinking about having a child, a genetic counselor can help you and your partner understand the implications of having a child.

You have other concerns about your results, consult with a healthcare professional.

Learn more about this condition and connect with support groups.

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**ABCBC8-related familial hyperinsulinism is caused by variants in the ABCBC8 gene.**

The ABCBC8 gene contains instructions for making a protein called sulfurylase, also known as ABCBC8. This protein is found in the pancreas and helps control the amount of insulin released into the bloodstream. Certain mutations in ABCBC8 can lead to an overproduction of insulin, which can cause low blood sugar levels.

Read more at [Genetics Home Reference](#)

### Test Indications for Use

The ABCBC8 test is used to identify defects in the ABCBC8 gene, which can cause familial hyperinsulinism. This test is not used to determine carrier status for ABCBC8-related familial hyperinsulinism in infants, but it can determine if a person has two copies of a variant. The report also lists if the result is associated with personal risk factors, such as family history or previous health problems.

#### Special Considerations

- Symptoms of familial hyperinsulinism may vary between people with the condition, even if they have the same genetic variant.
- There are no specific guidelines for this in the U.S. for carrier testing in family members. However, clinicians do have a testing protocol for familial hyperinsulinism. People with a family history of the condition may consider genetic counseling.

#### Test Performance Summary

- **Accuracy:** 95%
- **Compliance:** 95%
- **Sensitivity:** 95%
- **Specificity:** 95%
- **Test turnaround time:** 5 days
- **Test indication:** Genetic testing for familial hyperinsulinism.

### Warnings and Limitations

- This test does not diagnose health conditions.
- A positive result indicates that the condition may be present, but further testing is required to confirm the diagnosis.
- This test is not intended for use in non-clinical settings.
- There may be other factors to consider in evaluating the test results.

### References

**Frequently Asked Questions**

**ABCC2-related Familial Hypersensitivity**

This test looks for three genetic variants in the ABCC2 gene that are linked to familial hypersensitivity. People with two variants are at risk of developing symptoms of allergic sensitivity, and they will most likely pass a variant on to each of their children. People with one variant may have a small degree of hypersensitivity called familial hyperreactivity. They may also pass a variant on to their children. This test does not include all possible genetic variants associated with familial hyperreactivity.

Is this test useful? **Yes** | **No**

This test does not diagnose familial hyperreactivity. Only a healthcare professional can do that.

This test does not include all possible variants in the ABCC2 gene that are linked to familial hyperreactivity.

This test does not include variants in other genes such as H1FNG that are linked to familial hyperreactivity.

Is this test useful? **Yes** | **No**

**My report says my variant was detected. What does this mean?**

This means you have one of the genetic variants we tested, and you are at a risk for familial hyperreactivity. You can pass this variant on to each of your children. If your partner is a carrier for an ABCC2-related familial hyperreactivity, each of your children may have a 50% chance of having the condition.

Is this test useful? **Yes** | **No**

Most people with one variant do not develop familial hyperreactivity.

However, a small fraction of people with the variant may develop familial hyperreactivity, typically by early adulthood. One study in people of Ashkenazi Jewish descent estimated that about 1 in 100 people who inherit a single variant from their mother are not expected to develop familial hyperreactivity.

You may have a small degree of familial hyperreactivity, but no one in your family has been tested.

Is this test useful? **Yes** | **No**

**My report says my variant is something else. What does this mean?**

Based on your genetic result, you could pass a variant on to each of your children. If you’re starting a family, a genetic counselor can help you and your partner understand additional testing that might be appropriate.

In addition, a small fraction of people with your result may develop a form of hyperreactivity called familial hyperuricemia. Consider talking to a healthcare professional about your result.

You may also wish to share your results with your family. Because you share half of your mother’s genes, your genetic result could be relevant to your family members.

Is this test useful? **Yes** | **No**

How would this result affect my children?

Between you and one variant, you could pass this variant on to each of your children. If your partner is a carrier for ABCC2-related familial hyperreactivity, each of your children may have a 25% chance of having the condition.

For males with this result, if your partner is not a carrier, each child may still have a small chance of having the condition. One study in people of Ashkenazi Jewish descent estimated that about 1 in 100 people who inherit a single variant from their mother are not expected to develop familial hyperreactivity.

You may also wish to share your results with your family. Because you share half of your mother’s genes, your genetic result could be relevant to your family members.

Is this test useful? **Yes** | **No**

More questions? Check out our Customer Care Help Center.

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**Disclaimer:** The information provided is for educational purposes only. It should not be used as a substitute for professional medical advice. Always consult with a healthcare provider before making any significant changes to your health or medical regimen.